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ABSTRACT

This booklet presents facts about Down Syndrome for women over 35 years of age. Down Syndrome is one of the most common causes of severe mental retardation. The chances of bearing a child with a genetic defect such as Down Syndrome increase sharply with maternal age (1 in 900 for women aged 30 to 1 in 12 for women aged 48). Answers are provided to the following questions: (1) What is Down Syndrome? (2) How can an infant with Down Syndrome be identified? (3) What is the outlook for a child with Down Syndrome? (4) How retarded will a child with Down Syndrome be? (5) What causes Down Syndrome? (6) Why is Down Syndrome called a genetic defect? (7) What is the likelihood of having a child with Down Syndrome? (8) Why does the risk of having a baby with Down Syndrome increase with the mother's age? (9) What can I do to have a healthy baby? and (10) Does a genetic counselor recommend that you don't have children? (CB)

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facts about down syndrome for women over 35

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Much remains to be learned about genetic defects. The National Institute of Child Health and Human Development is one of many organizations concerned with genetic diseases. The Institute supports and conducts research aimed at improving each individual's growth and development and providing new and better methods of prevention, detection, and treatment of diseases, particularly those affecting women and children.

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introduction

Every pregnant woman wants to have a healthy baby. If you are planning a pregnancy and are over 35 years old, there are some facts you should know about **Down syndrome (mongolism).***

The chances of bearing a child with a **genetic defect** such as Down syndrome increase sharply for women over 35, and even more for those over 40.

If you are considering having children at a later time or if you have a child with Down syndrome in your family, this pamphlet contains answers to questions of concern to you.

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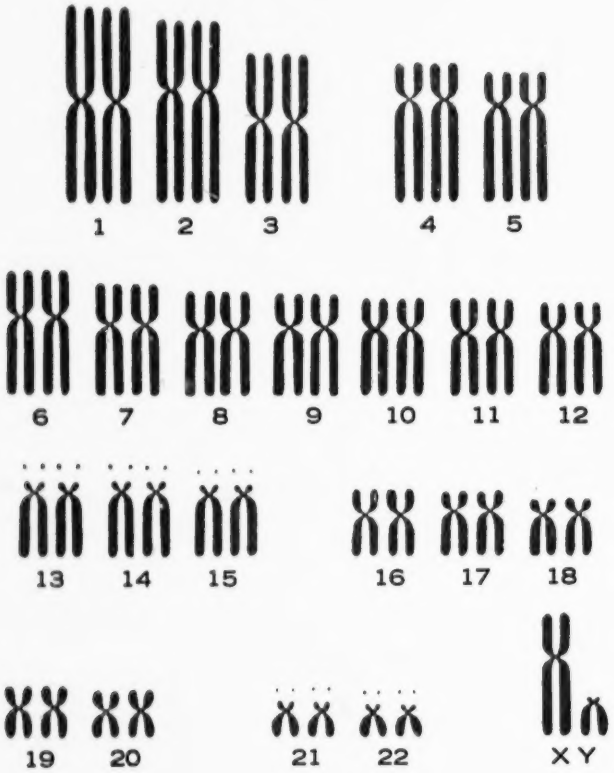
* Words appearing in **bold** are explained in the Glossary.

Normal Karyotype



All normal cells (except ova and sperm) have 46 chromosomes—44 autosomes and 2 sex chromosomes. Scientists have given these chromosomes numbers and have paired them according to their similarities. (Note the sex chromosome: all normal males have one "X" and one "Y" chromosome; normal females have two "X" chromosomes.)

Q. What is Down syndrome?

A. Down syndrome is one of the most common causes of severe mental retardation. It is characterized by various physical and mental abnormalities and by the presence of an extra **chromosome.**

Q. How can an infant with Down syndrome be identified?

A. In examining a newborn thought to have Down syndrome, the doctor will usually find certain telltale characteristics—slanting eyes, slightly protruding lips, small ears, slightly protruding tongue, short hands, feet, and trunk, and sometimes, an unusual crease in the palm of the baby's hand.

Q. What is the outlook for a child with Down syndrome?

A. The most serious problem of children with Down syndrome is mental retardation. A baby with Down syndrome is often slow in learning to turn over, sit up, crawl, walk, and speak. In addition, he almost never achieves near normal intelligence.

Also, babies with Down syndrome have more heart defects as well as more frequent colds and respiratory infections than the normal infant.

Since the advent of antibiotics and modern heart surgery, the life expectancy of a child with Down syndrome has greatly increased. For those with Down syndrome who survive the first few years, death rates are about the same as for normal persons until around age 40. Because people with Down syndrome seem to age a little more rapidly than normal, they sometimes become susceptible to diseases associated with old age as early as age 40.

Society is becoming more understanding of mental retardation and has set up more educational and vocational training programs for mentally retarded individuals. At this time, there is no cure for Down syndrome. However, if parents can accept the initial setback of learning that their child is abnormal, they can provide good, loving homes as well as derive satisfaction from their child.

Q. How retarded will a child with Down syndrome be?

A. Generally, children with Down syndrome have IQs in the 30s to 50s, occasionally in the 60s and 70s. (Average scores for normal children are in the range of 90-110.) Studies by some scientists have shown that children with Down syndrome who have been institutionalized have lower IQ scores than those who have remained at home. One explanation for this is that a child reared at home receives more attention and greater stimulation than one brought up in an institution. There are, however, special occasions when institutional care is in the best interests of the child and family.

Q. What causes Down syndrome?

A. The cause of Down syndrome is unknown, but it is reassuring to know that nothing a mother or father does during pregnancy can cause it.

Q. Why is Down syndrome called a genetic defect?

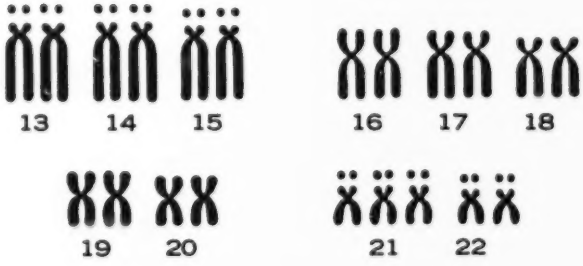
A. The child with Down syndrome has extra genetic material, usually in the form of an entire extra chromosome. The extra chromosome can come from **either** the mother or the father. The event which results in this "extra" chromosome usually occurs during development of the **egg** or **sperm**

or during **fertilization.** The extra chromosome resembles two other chromosomes designated by scientists, "pair 21." Although other genetic diseases may be associated with an extra chromosome, only Down syndrome is characterized by extra "chromosome 21" material.

There are three genetic types of Down syndrome:

1. **Trisomy 21:** This condition occurs in about 95 percent of all cases of Down syndrome. Though his parents have normal chromosomes, the child with trisomy Down syndrome has three, rather than the normal two chromosomes in pair 21. Trisomy 21 usually results from a mistake in cell division of either the sperm or egg and is an accidental occurrence. The incidence of trisomy 21 increases dramatically with the age of the mother.

Trisomy 21

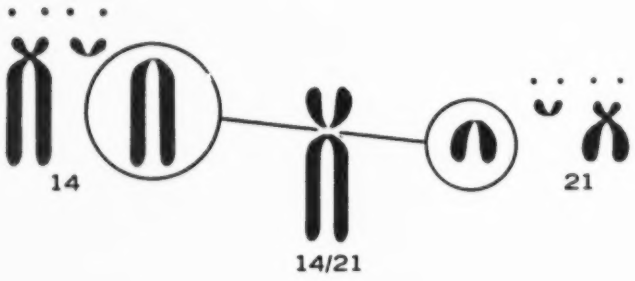


Trisomy 21 refers to a condition in which there are three rather than the usual pair of chromosomes in the "21" pair. Trisomy 21 usually results from a mistake in cell division of either the egg or the sperm and is an accidental occurrence.

2. **Translocation:** This genetic condition can take several forms, and it occurs in about four percent of Down syndrome cases. Children with **translocation Down syndrome** have an extra number 21 chromosome which has broken and become attached to another chromosome. A parent can carry a translocation without showing any symptoms of disease because the parent still carries the correct amount of genetic material, although some of it is out of place (translocated).

Normal children receive only one chromosome of pair 21 from each parent. But a parent with a translocation can pass on his or her normal chromosome 21 plus the translocated chromosome 21, giving the child too much chromosome 21 material.

Formation of a Translocation Chromosome



A translocation occurs when a piece of one chromosome breaks off and attaches to a different chromosome. This "translocation chromosome" has been formed by the breaking and rejoining of chromosomes 14 and 21 (with loss of the little pieces).

3. **Mosaicism:** This genetic type of Down syndrome is very rare. It occurs when an individual has cells with different chromosome counts (for example, 46 in some cells and 47 in others). Mosaicism is not carried in the parents' chromosomes; it is accidental, resulting from an error which occurs in the early cell division of the fertilized egg. Babies with mosaic Down syndrome may have only some of the features of the syndrome, because only some of their cells have an abnormal number of chromosomes.

Q. What is the likelihood of having a child with Down syndrome?

A. The chances of bearing a baby with Down syndrome rise with maternal age. This rise increases sharply after age 40. Although only 4 percent of all pregnancies in this country occur in women over 35, roughly one fourth of the babies with Down syndrome are born to women in this age group. The chance of bearing a baby with Down syndrome when you're age 35 is one in 400; at age 40 the chance is one in 100; at age 44 the chance is one in 35; and at 48 years of age the chance rises to one in 12. By contrast, women under 30 years old have less than one chance in 1000 of having a child with Down syndrome.

Q. Why does the risk of having a baby with Down syndrome increase with the mother's age?

A. Scientists have several theories to explain the increased risk. Women are born with all the eggs which are necessary for bearing children. By the time a woman is age 35, her eggs are also 35 years old. Some scientists believe that just as

Relationship of Down Syndrome to Maternal Age

Mother's Age	Incidence of Down Syndrome
under 30	less than 1 in 1,000
30	1 in 900
35	1 in 400
36	1 in 300
37	1 in 230
38	1 in 180
39	1 in 135
40	1 in 105
42	1 in 60
44	1 in 35
46	1 in 20
48	1 in 12

The risk of having a child with Down syndrome increases with maternal age.

people pass their prime in certain bodily functions, so may eggs.

Also, because the egg of a woman over 35 has been in the ovary for that long a time, some scientists feel the egg is more likely to have been exposed to physical or chemical forces such as radiation which might damage the genetic material in the egg.

Q. What can I do to have a healthy baby?

A. Take good care of your own health, even before you have children. Employ **family planning**—that is, use birth control methods so that you have only the number of children you want, when you want them. See a physician as soon as you suspect you're pregnant and follow the diet and other guidelines he sets up for you.

Consult your family physician, gynecologist, or public health nurse IF:

- You are over age 35, OR
- you are already the parent of one child with Down syndrome or other birth defects, OR
- children with birth defects, genetic diseases or mental retardation have been born in your family or your husband's family, OR
- you have had great difficulty in becoming pregnant or have had several miscarriages.

The doctor or health professional you consult will assess whether it is advisable for you and your husband to see a **genetic counselor.**

Q. Does a genetic counselor recommend that you don't have children?

A. No, this a personal decision. A genetic counselor will only give you the information you need to evaluate the risk you might be facing. He may recommend that you and your husband have your chromosomes analyzed **(karyotyped)**. On the basis of this analysis, he can in some cases predict the chances of your having a healthy baby. For example, if the karyotype shows that the woman has a translocation commonly associated with Down syndrome, the genetic counselor would be able to predict the probabilities of her child being: 1) normal; 2) normal but carrying a translocation like the mother's; or 3) a victim of Down syndrome. The purpose of the genetic counselor is to help you understand the risk you face, so that you can plan your family thoughtfully.

If you are already pregnant, under certain circumstances your doctor or genetic counselor may suggest you undergo **amniocentesis** to find out whether the **fetus** which you are carrying has Down syndrome. Amniocentesis is a technique used by specially trained physicians to obtain amniotic fluid for analysis. If your doctor thinks you should have this test done, do NOT put it off. Amniocentesis is best performed between the 15th and 17th week after the first day of the last menstrual period; analysis of the amniotic fluid takes several weeks.

GLOSSARY

Amniocentesis—A medical procedure whereby fluid is drawn from the liquid-filled sac surrounding the developing fetus while in the womb. Cells shed from the fetus into the fluid can then be specially prepared within 2-3 weeks for chromosomal analysis. (See karyotype.)

Autosomes—22 of the 23 chromosomes normally contained in ova and sperm (reproductive) cells in the human body. In all other (non-reproductive) body cells, the chromosomes occur in pairs, so each cell has 46. Each autosome usually appears identical to its partner, but each pair is different in its genetic content and frequently in its appearance from all other pairs. The remaining pair of chromosomes are the sex chromosomes and are designated XX in females and XY in males.

Chromosomes—Chromosomes are structures which are present in every human cell and are composed of genes, which carry hereditary traits. All normal cells in the human body, except ova and sperm cells, have 46 chromosomes —44 autosomes and 2 sex chromosomes. Normal reproductive cells (ova and sperm) contain 23 chromosomes. (See autosomes and sex chromosomes.)

Down Syndrome—One of the most common causes of mental retardation. A combination of physical deformities and mental retardation, it is characterized by a genetic defect in chromosome pair 21. (See trisomy 21, translocation Down syndrome, and mosaicism.)

Egg—(See ovum.)

Family Planning—Family planning is the use of birth control methods in planning the number and timing of pregnancies.

Fertilization—Fertilization is the fusion of an egg from the woman and a sperm from the man, which then results in the start of the development of the offspring.

Fetus—The fetus is the unborn offspring. Before eleven weeks it is called an embryo. It is known as an infant or baby after it is born.

Gene—A gene is the smallest particle of hereditary information which is passed from parent to offspring. A group of genes together in a specific sequence make up the structures known as chromosomes.

Genetics—Genetics is the study of heredity or how traits (physical and mental) are passed from parents to offspring.

Genetic Counselor—A genetic counselor is an advisor who is usually qualified in several medical specialties such as internal medicine, pediatrics and genetics. The genetic counselor will compile a complete "family tree" of information about the genetic and general medical history of the patient and family. This history, together with appropriate laboratory tests, will help the genetic counselor predict the likelihood that genetic defects will occur in a family.

Genetic Defect—An abnormality of the genes. (See trisomy, translocation.)

Hereditary—Referring to traits passed from generation to generation via the genes and chromosomes.

Karyotyping—A charting system devised by scientists for numbering and pairing chromosomes on the basis of similar characteristics such as size and shape. A karyotype is made by photographing the chromosomes, cutting out their pictures and arranging them by pairs in a standard order. Karyotyping makes chromosomes easier to study.

Mongolism—Formerly an alternate name for Down syndrome, but no longer favored because of its negative connotations.

Mosaicism—Mosaicism refers to a genetic condition in which a person's genes are different in different cells of his body. A mosaic individual can pass on normal genes or abnormal genes to the offspring.

Ovulation—Release of the egg from the ovary. (See ovum.) In most women one egg is ovulated each month.

Ovum (plural: ova)—The ova are the female egg cells which are contained in the organs known as the ovaries. Reproduction is accomplished by the successful fertilization of the ovum with a sperm and the subsequent development of the fertilized ovum into a baby. Ova (and sperm) are unlike other cells in the human body because they each have only 23 chromosomes. When they join they then can produce an offspring with the correct number, 46 chromosomes.

Sex Chromosomes—The sex chromosomes are the two chromosomes designated "X" and "Y" by the scientists. All normal males have one "Y" and one "X" chromosome. The normal female has two "X" chromosomes and no "Y" chromosomes. The other 44 chromosomes in the normal cell are known as autosomes.

Sperm—The male reproductive cells. (See ovum.)

Translocation—The transfer of a piece of one chromosome to a different chromosome.

Translocation Down Syndrome—A form of Down syndrome which may be hereditary and which results from a piece of a chromosome in pair 21 breaking off and attaching itself to another chromosome.

Trisomy—Trisomy refers to a condition in which there are three, rather than the usual pair of chromosomes at a designated place in the karyotype.

Trisomy 21—Trisomy 21 refers to the presence of three chromosomes rather than the normal pair designated 21. This genetic defect is the chromosomal abnormality most frequently associated with Down syndrome. Trisomy 21 is sometimes used as an alternate name for Down syndrome.

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